OTTO VON GUERICKE UNIVERSITÄT MAGDEBURG

MED MEDIZINISCHE FAKULTÄT

Forschungsbericht 2018

Institut für Humangenetik

INSTITUT FÜR HUMANGENETIK

Leipziger Str. 44, 39120 Magdeburg
Tel. 49 (0)391 67 15062, Fax 49 (0)391 67 15066
martin.zenker@med.ovgu.de

1. Leitung

Prof. Dr. Martin Zenker

2. HochschullehrerInnen

PD Dr. Ilse Wieland

3. Forschungsprofil

Neurogenetik

- X-chromosomal erbliche geistige Retardierung
- Molekulargenetische Analyse neuromuskulärer Erkrankungen

Tumorgenetik

- Zytogenetische und molekularzytogenetische Charakterisierung von Leukämien
- Genkartierung ausgewählter genetischer Syndrome
- Molekulargenetische Analyse der ursache bei familiärer Spalthand-/Spaltfussfehlbildungen

4. Kooperationen

- Universitätsmedizin Greifswald

5 Veröffentlichungen

Begutachtete Zeitschriftenaufsaetze

Asadollahi, Reza; Strauss, Justin E.; Zenker, Martin; Beuing, Oliver; Edvardson, Simon; Elpeleg, Orly; Strom, Tim M.; Joset, Pascal; Niedrist, Dunja; Otte, Christine; Oneda, Beatrice; Boonsawat, Paranchai; Azzarello-Burri, Silvia; Bartholdi, Deborah; Papik, Michael; Zweier, Markus; Haas, Cordula; Ekici, Arif Bülent; Baumer, Alessandra; Boltshauser, Eugen; Steindl, Katharina; Nothnagel, Michael; Schinzel, Albert; Stoeckli, Esther; Rauch, Anita
Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling
European journal of human genetics: the official journal of the European Society of Human Genetics - Basingstoke: Stockton Press, Bd. 26.2018, 2, S. 197-209;
[Imp.fact.: 3.636]

Ashraf, Shazia; Kudo, Hiroki; Rao, Jia; Kikuchi, Atsuo; Widmeier, Eugen; Lawson, Jennifer A.; Tan, Weizhen; Hermle, Tobias Franz; Warejko, Jillian K.; Shril, Shirlee; Airik, Merlin; Jobst-Schwan, Tilman; Lovric, Svjetlana; Braun, Daniela A.; Gee, Heon Yung; Schapiro, David; Majmundar, Amar J.; Sadowski, Carolin E.; Pabst, Werner L.; Daga, Ankana; Ven, Amelie; Schmidt, Johanna M.; Low, Boon Chuan; Gupta, Anjali Bansal; Tripathi, Brajendra K.; Wong, Jenny; Campbell, Kirk; Metcalfe, Kay; Schanze, Denny; Niihori, Tetsuya; Kaito, Hiroshi; Nozu, Kandai; Tsukaguchi, Hiroyasu; Tanaka, Ryojiro; Hamahira, Kiyoshi; Kobayashi, Yasuko; Takizawa, Takumi; Funayama, Ryo; Nakayama, Keiko; Aoki, Yoko; Kumagai, Naonori; Iijima, Kazumoto; Fehrenbach, Henry; Kari, Jameela A.; Desoky, Sherif; Jalalah, Sawsan; Bogdanovic, Radovan; Staji, Nataa; Zappel, Hildegard; Rakhmetova, Assel; Wassmer, Sharon-Rose; Jungraithmayr, Therese; Strehlau, Jürgen; Kumar, Aravind Selvin; Bagga, Arvind; Soliman, Neveen A.; Mane, Shrikant M.; Kaufman, Lewis; Lowy, Douglas R.; Jairajpuri, Mohamad A.; Lifton, Richard P.; Pei, York; Zenker, Martin; Kure, Shigeo; Hildebrandt, Friedhelm
Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment
Nature Communications - [London]: Nature Publishing Group UK, Bd. 9.2018, Art.-Nr. 1960, insges. 14 S.;
[Imp.fact.: 12.353]

Beaufils, Camille; Farlay, Delphine; Machuca-Gayet, Irma; Fassier, Alice; Zenker, Martin; Freychet, Caroline; Bonnelye, Edith; Bertholet-Thomas, Aurélia; Ranchin, Bruno; Bacchetta, Justine
Skeletal impairment in Pierson syndrome - is there a role for laminin[beta]2 in bone physiology?
Bone: cell molecular biology, pathophysiology, treatment : official journal of the International Bone and Mineral Society - Amsterdam [u.a.]: Elsevier Science, Bd. 106.2018, S. 187-193;
[Imp.fact.: 4.455]

Dahlmann, Julia; Awad, George; Dolny, Carsten; Weinert, Sönke; Richter, Karin; Fischer, Klaus-Dieter; Munsch, Thomas; Leßmann, Volkmar; Volleth, Marianne; Zenker, Martin; Chen, Yaoyao; Merkl, Claudia; Schnieke, Angelika; Baraki, Hassina; Kutschka, Ingo; Kensah, George
Generation of functional cardiomyocytes from rat embryonic and induced pluripotent stem cells using feeder-free expansion and differentiation in suspension culture
PLOS ONE - San Francisco, California, US: PLOS, Bd. 13.2018, 3, Art.-Nr. e0192652, insges. 22 S.;
[Imp.fact.: 2.766]

Grant, Andrew R.; Cushman, Brandon J.; Cavé, Hélène; Dillon, Mitchell W.; Gelb, Bruce D.; Gripp, Karen W.; Lee, Jennifer A.; Mason-Suares, Heather; Rauen, Katherine A.; Tartaglia, Marco; Vincent, Lisa M.; Zenker, Martin
Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework
Human mutation - New York, NY [u.a.]: Wiley-Liss, Bd. 39.2018, 11, S. 1485-1493;
[Imp.fact.: 5.359]

Haghighi, Fereshteh; Dahlmann, Julia; Nakhaei-Rad, Saeideh; Lang, Alexander; Kutschka, Ingo; Zenker, Martin; Kensah, George; Piekorz, Roland; Ahmadian, Mohammad Reza
bFGF-mediated pluripotency maintenance in human induced pluripotent stem cells is associated with NRAS-MAPK signaling
Cell communication and signaling - London: Biomed Central, Bd. 16.2018, Art.-Nr. 96, insges. 14 S.;
[Imp.fact.: 5.324]

Harms, Frederike L.; Alawi, Malik; Amor, David J.; Tan, Tiong Y.; Cuturilo, Goran; Lißewski, Christina; Brinkmann, Julia; Schanze, Denny; Kutsche, Kerstin; Zenker, Martin
The novel RAF1 mutation p.(Gly361Ala) located outside the kinase domain of the CR3 region in two patients with Noonan syndrome, including one with a rare brain tumor
American journal of medical genetics / A - New York, NY: Wiley-Liss, Bd. 176.2018, 2, S. 470-476;
[Imp.fact.: 2.264]

Hauer, Nadine N.; Popp, Bernt; Schoeller, Eva; Schuhmann, Sarah; Heath, Karen E.; Hisado-Oliva, Alfonso; Klinger, Patricia; Kraus, Cornelia; Trautmann, Udo; Zenker, Martin; Zweier, Christiane; Wiesener, Antje; Abou Jamra, Rami; Kunstmann, Erdmute; Wieczorek, Dagmar; Uebe, Steffen; Ferrazzi, Fulvia; Büttner, Christian; Ekici, Arif Bülent; Rauch, Anita; Sticht, Heinrich; Dörr, Helmuth-Günther; Reis, André; Thiel, Christian T.
Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature
Genetics in medicine: official journal of the American College of Medical Genetics - London, UK: Springer Nature, Bd. 20.2018, 6, S. 630-638;
[Imp.fact.: 9.937]

Ivanovski, Ivan; Djuric, Olivera; Caraffi, Stefano Giuseppe; Santodirocco, Daniela; Pollazzon, Marzia; Rosato, Simonetta; Cordelli, Duccio Maria; Abdalla, Ebtesam; Accorsi, Patrizia; Adam, Margaret P.; Ajmone, Paola Francesca; Badura-Stronka, Magdalena; Baldo, Chiara; Baldi, Maddalena; Bayat, Allan; Bigoni, Stefania; Bonvicini, Federico; Breckpot, Jeroen; Callewaert, Bert; Cocchi, Guido; Cuturilo, Goran; De Brasi, Daniele; Devriendt, Koenraad; Dinulos, Mary Beth; HjortshØj, Tina Duelund; Epifanio, Roberta; Faravelli, Francesca; Fiumara, Agata; Formisano, Debora; Giordano, Lucio; Grasso, Marina; GrØnborg, Sabine; Iodice, Alessandro; Iughetti, Lorenzo; Kuburovic, Vladimir; Kutkowska-Kazmierczak, Anna; Lacombe, Didier; Lo Rizzo, Caterine; Luchetti, Anna; Malbora, Baris; Mammi, Isabella; Mari, Francesca; Montorsi, Giulia; Moutton, Sebastien; Möller, Rikke Steensbjerre; Muschke, Petra; Nielsen, Jens Erik Klint; Obersztyn, Ewa; Pantaleoni, Chiara; Pellicciari, Alessandro; Pisanti, Maria Antonietta; Prpic, Igor; Poch-Olive, Maria Luisa; Raviglione, Federico; Renieri, Alessandra; Ricci, Emilia; Rivieri, Francesca; Santen, Gijs W.; Savasta, Salvatore; Scarano, Gioacchino; Schanze, Ina; Selicorni, Angelo; Silengo, Margherita; Smigiel, Robert; Spaccini, Luigina; Sorge, Giovanni; Szczaluba, Krzysztof; Tarani, Luigi; Tone, Luis Gonzaga; Toutain, Annick; Trimouille, Aurelien; Valera, Elvis Terci; Vergano, Samantha Schrier; Zanotta, Nicoletta; Zenker, Martin; Conidi, Andrea; Zollino, Marcella; Rauch, Anita; Zweier, Christiane; Garavelli, Livia
Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care
Genetics in medicine: official journal of the American College of Medical Genetics - London, UK: Springer Nature, Bd. 20.2018, 9, S. 965-975;
[Imp.fact.: 9.937]

Johnston, Jennifer J.; Smagt, Jasper J.; Rosenfeld, Jill A.; Pagnamenta, Alistar T.; Alswaid, Abdulrahman; Baker, Eva H.; Blair, Edward; Borck, Guntram; Brinkmann, Julia; Craigen, William; Dung, Vu Chi; Emrick, Lisa; Everman, David B.; Gassen, Koen L.; Gulsuner, Suleyman; Harr, Margaret H.; Jain, Mahim; Küchler, Alma; Leppig, Kathleen A.; McDonald-McGinn, Donna M.; Can, Ngoc Thi Bich; Peleg, Amir; Roeder, Elizabeth R.; Rogers, R. Curtis; Sagi-Dain, Lena; Sapp, Julie C.; Schäffer, Alejandro A.; Schanze, Denny; Stewart, Helen; Taylor, Jenny C.; Verbeek, Nienke E.; Walkiewicz, Magdalena A.; Zackai, Elaine H.; Zweier, Christiane; Zenker, Martin; Lee, Brendan; Biesecker, Leslie G.
Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants
Genetics in medicine: official journal of the American College of Medical Genetics - London, UK: Springer Nature, Bd. 20.2018, 10, S. 1175-1185;
[Imp.fact.: 9.937]

Link, Jastin; Thon, Cosima; Schanze, Denny; Steponaitiene, Ruta; Kupinskas, Juozas; Zenker, Martin; Canbay, Ali E.; Malfertheiner, Peter; Link, Alexander
Food-derived xeno-microRNAs - influence of diet and detectability in gastrointestinal tract : proof-of-principle study
Molecular nutrition & food research - Weinheim: Wiley-VCH, Bd. 62.2018;
[Imp.fact.: 5.151]

Martinelli, Simone; Krumbach, Oliver H. F.; Pantaleoni, Francesca; Coppola, Simona; Amin, Ehsan; Pannone, Luca; Nouri, Kazem; Farina, Luciapia; Dvorsky, Radovan; Lepri, Francesca; Buchholzer, Marcel; Konopatzki, Raphael; Walsh, Laurence; Payne, Katelyn; Pierpont, Mary Ella; Vergano, Samantha Schrier; Langley, Katherine G.; Larsen, Douglas; Farwell, Kelly D.; Tang, Sha; Mroske, Cameron; Gallotta, Ivan; Schiavi, Elia; Monica, Matteo; Lugli, Licia; Rossi, Cesare; Seri, Marco; Cocchi, Guido; Henderson, Lindsay; Baskin, Berivan; Alders, Marielle; Mendoza-Londono, Roberto; Dupuis, Lucie; Nickerson, Deborah A.; Chong, Jessica X.; Meeks, Naomi; Brown, Kathleen; Causey, Tahnee; Cho, Megan T.; Demuth, Stephanie; Digilio, Maria Cristina; Gelb, Bruce D.; Bamshad, Michael J.; Zenker, Martin; Ahmadian, Mohammad Reza; Hennekam, Raoul C.; Tartaglia, Marco; Mirzaa, Ghayda M.
Functional dysregulation of CDC42 causes diverse developmental phenotypes
The American journal of human genetics - New York, NY [u.a.]: Cell Press, Bd. 102.2018, 2, S. 309-320;
[Imp.fact.: 8.855]

Meester, Josephina A. N.; Sukalo, Maja; Schröder, Kim C.; Schanze, Denny; Baynam, Gareth; Borck, Guntram; Brämswig, Nuria; Duman, Duygu; Gilbert-Dussardier, Brigitte; Holder-Espinasse, Muriel; Itin, Peter; Johnson, Diana S.; Joss, Shelagh; Koillinen, Hannele; McKenzie, Fiona; Morton, Jenny; Nelle, Heike Maria; Reardon, Willie; Roll, Claudia; Salih, Mustafa Abdalla M.; Savarirayan, Ravi; Scurr, Ingrid; Splitt, Miranda; Thompson, Elizabeth; Titheradge, Hannah; Travers, Colm P.; Maldergem, Lionel; Whiteford, Margo; Wieczorek, Dagmar; Vandeweyer, Geert; Trembath, Richard; Laer, Lut; Loeys, Bart L.; Zenker, Martin; Southgate, Laura; Wuyts, Wim
Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort
Human mutation - New York, NY [u.a.]: Wiley-Liss, Bd. 39.2018, 9, S. 1246-1261;
[Imp.fact.: 5.359]

Meyer zum Büschenfelde, Uta; Brandenstein, Laura Isabel; Elsner, Leonie; Flato, Kristina; Holling, Tess; Zenker, Martin; Rosenberger, Georg; Kutsche, Kerstin
RIT1 controls actin dynamics via complex formation with RAC1/CDC42 and PAK1
PLoS Genetics: a peer-reviewed, open-access journal - San Francisco, Calif: Public Library of Science, Bd. 14.2018, 5, Art.-Nr. e1007370, insges. 28 S.;
[Imp.fact.: 5.54]

Neusel, Chantal; Class, Dieter; Eckert, Alexander W.; Firsching, Raimund; Göbel, Peter; Götz, Dorit; Haase, Roland; Jorch, Gerhard; Köhn, Andrea; Kropf, Siegfried; Patzer, Ludwig; Schanze, Ina; Zahl, Christian; Rißmann, Anke
Multicentre approach to epidemiological aspects of craniosynostosis in Germany
The British journal of oral & maxillofacial surgery - Amsterdam [u.a.]: Elsevier, Bd. 56.2018, 9, S. 881-886;
[Imp.fact.: 1.26]

Priolo, Manuela; Schanze, Denny; TattonBrown, Katrin; Mulder, Paul A.; Tenorio, Jair; Kooblall, Kreepa; Acero, Inés Hernández; Alkuraya, Fowzan S.; Arias, Pedro; Bernardini, Laura; Bijlsma, Emilia K.; Cole, Trevor; Coubes, Christine; Dapia, Irene; Davies, Sally; Donato, Nataliya Di; Elcioglu, Nursel H.; Fahrner, Jill A.; Foster, Alison; González, Noelia García; Huber, Ilka; Iascone, Maria; Kaiser, Ann-Sophie; Kamath, Arveen; Liebelt, Jan; Lynch, Sally Ann; Maas, Saskia M.; Mammì, Corrado; Mathijssen, Inge B.; McKee, Shane; Menke, Leonie A.; Mirzaa, Ghayda M.; Montgomery, Tara; Neubauer, Dorothee; Neumann, Thomas E.; Pintomalli, Letizia; Pisanti, Maria Antonietta; Plomp, Astrid S.; Price, Sue; Salter, Claire; SantosSimarro, Fernando; Sarda, Pierre; Segovia, Mabel; ShawSmith, Charles; Smithson, Sarah; Suri, Mohnish; Valdez, Rita Maria; Haeringen, Arie; Hagen, Johanna M.; Zollino, Marcela; Lapunzina, Pablo; Thakker, Rajesh V.; Zenker, Martin; Hennekam, Raoul C.
Further delineation of Malan syndrome
Human mutation - New York, NY [u.a.]: Wiley-Liss, Bd. 39.2018, 9, S. 1226-1237;
[Imp.fact.: 5.359]

Schanze, Ina; Bunt, Jens; Lim, Jonathan W. C.; Schanze, Denny; Dean, Ryan J.; Alders, Marielle; Blanchet, Patricia; Attié-Bitach, Tania; Berland, Siren; Boogert, Steven; Boppudi, Sangamitra; Bridges, Caitlin J.; Cho, Megan T.; Dobyns, William B.; Donnai, Dian; Douglas, Jessica; Earl, Dawn L.; Edwards, Timothy J.; Faivre, Laurence; Fregeau, Brieana; Genevieve, David; Gérard, Marion; Gatinois, Vincent; Holder-Espinasse, Muriel; Huth, Samuel F.; Izumi, Kosuke; Kerr, Bronwyn; Lacaze, Elodie; Lakeman, Phillis; Mahida, Sonal; Mirzaa, Ghayda M.; Morgan, Sian M.; Nowak, Catherine; Peeters, Hilde; Petit, Florence; Pilz, Daniela; Puechberty, Jacques; Reinstein, Eyal; Rivière, Jean-Baptiste;

Santani, Avni B.; Schneider, Anouck; Sherr, Elliott H.; Smith-Hicks, Constance; Wieland, Ilse; Zackai, Elaine; Zhao, Xiaonan; Gronostajski, Richard M.; Zenker, Martin; Richards, Linda J.
NFIB haploinsufficiency is associated with intellectual disability and macrocephaly
The American journal of human genetics - New York, NY [u.a.]: Cell Press, Bd. 103.2018, 5, S. 752-768;
[Imp.fact.: 8.855]

Schiefer, Jan; Zenker, Martin; Gröne, Hermann-Josef; Chatzikyrkou, Christos; Mertens, Peter Rene; Liakopoulos, Vassilios
Unrecognized juvenile nephropathic cystinosis
Kidney international: official journal of the International Society of Nephrology - New York, NY: Elsevier, Bd. 94.2018, 5, S. 1027;
[Imp.fact.: 8.429]

Ufartes, Roser; Schwenty-Lara, Janina; Freese, Luisa; Neuhofer, Christiane Michaela; Möller, Janika; Wehner, Peter; Revenswaaij-Arts, Conny M. A.; Wong, Monica T. Y.; Schanze, Ina; Tzschach, Andreas; Bartsch, Oliver; Borchers, Annette; Pauli, Silke Jeannette
Sema3a plays a role in the pathogenesis of CHARGE syndrome
Human molecular genetics: HMG online - Oxford: Oxford Univ. Press, Bd. 27.2018, 8, S. 1343-1352;
[Imp.fact.: 4.902]

Zenker, Martin; Tinschert, Sigrid; Wieland, Ilse; Schanze, Denny; Happle, Rudolf
A postzygotic SMO mutation caused the original case of HappleTinschert syndrome
Acta dermato-venereologica: a journal for clinical and experimental research in the field of dermatology and venereology - Uppsala: Acta Dermato-Venereologica, Bd. 98.2018, 5, S. 534-535;
[Imp.fact.: 3.127]

Abstracts

Steinbusch, Mandy M.; Caron, Marjolein M.; Surtel, Don A.; Dijk, Paul J.; Friedrich, Franziska; Zabel, Bernhard; Rhijn, Lodewijk W.; Peffers, Mandy J.; Welting, Tim J.
Viperin regulates chondrogenic differentiation via CXCL10 protein secretion
Osteoarthritis and cartilage - [S.l.]: Elsevier, Bd. 26.2018, Suppl. 1, S. S93;
[Imp.fact.: 5.454]

Dissertationen

Becker, Jana; Wieland, Ilse [GutachterIn]; Wieczorek, Dagmar [GutachterIn]
Retrospektive Charakterisierung von Patienten mit mentaler Retardierung/Intelligenzminderung aus dem Institut für Humangenetik des Universitätsklinikums Magdeburg und molekulargenetische Aufarbeitung ausgewählter familiärer Fälle
Magdeburg: Otto-von-Guericke-Universität Magdeburg, 2018, VI, 92 Blätter, Illustrationen, Diagramme

Boppudi, Sangamitra; Zenker, Martin [GutachterIn]
About the impact of altered RAS-MAPK and PI3K-AKT signalling in human developmental disorders
Magdeburg, 2017, IV, 196 Seiten, Illustrationen, Tabellen, Diagramme;
[Name des Verfassers auf dem Umschlag: Boppudi, Sanga Mitra; Literaturverzeichnis: Seite 138-149]

Lißewski, Christina; Zenker, Martin [GutachterIn]
The RASopathies - molecular genetics and genotype-phenotype correlations
Magdeburg, 2017, 178 Blätter, Illustrationen, Diagramme, 30 cm;
[Literaturverzeichnis: Blatt 134-144]